



## PDGFRB gene

platelet derived growth factor receptor beta

### Normal Function

The *PDGFRB* gene provides instructions for making a protein called platelet-derived growth factor receptor beta (PDGFR $\beta$ ), which is part of a family of proteins called receptor tyrosine kinases. Receptor tyrosine kinases transmit signals from the cell surface into the cell through a process called signal transduction. The PDGFR $\beta$  protein is found in the cell membrane of certain cell types, where a protein called platelet-derived growth factor attaches (binds) to it. This binding turns on (activates) the PDGFR $\beta$  protein, which then activates other proteins inside the cell by adding a cluster of oxygen and phosphorus atoms (a phosphate group) at specific positions. This process, called phosphorylation, leads to the activation of a series of proteins in multiple signaling pathways.

The signaling pathways stimulated by the PDGFR $\beta$  protein control many important processes in the cell such as growth and division (proliferation), movement, and survival. PDGFR $\beta$  protein signaling is important for the development of many types of cells throughout the body.

### Health Conditions Related to Genetic Changes

#### familial idiopathic basal ganglia calcification

At least two mutations in the *PDGFRB* gene have been found to cause familial idiopathic basal ganglia calcification (FIBGC). This condition is characterized by abnormal deposits of calcium (calcification) in the brain, which can lead to movement and psychiatric problems. These mutations change single protein building blocks (amino acids) in the PDGFR $\beta$  protein. It is unclear how *PDGFRB* gene mutations cause FIBGC. Mutations may alter signaling within cells that line blood vessels in the brain, causing them to take in excess calcium, and leading to calcification of the lining of these blood vessels. Alternatively, changes in the PDGFR $\beta$  protein can alter phosphate transport signaling pathways, causing an increase in phosphate levels. In the brain, the excess phosphate combines with calcium and forms deposits.

The *PDGFRB* gene is active (expressed) throughout the body; it is unclear why the effects of these mutations are limited to the basal ganglia and other brain regions that are involved in FIBGC.

## PDGFRB-associated chronic eosinophilic leukemia

Genetic rearrangements (translocations) involving the *PDGFRB* gene cause a type of cancer of blood-forming cells called *PDGFRB*-associated chronic eosinophilic leukemia. This condition is characterized by an increased number of eosinophils, a type of white blood cell. The most common of these translocations brings together part of the *PDGFRB* gene with another gene called *ETV6*, whose function is to turn off gene activity. Together, these pieces create the *ETV6-PDGFRB* fusion gene. Occasionally, genes other than *ETV6* are fused with the *PDGFRB* gene. The translocations that lead to these fusion genes are somatic mutations, which are acquired during a person's lifetime and occur initially in a single cell. This cell continues to grow and divide, producing a group of cells with the same mutation (a clonal population).

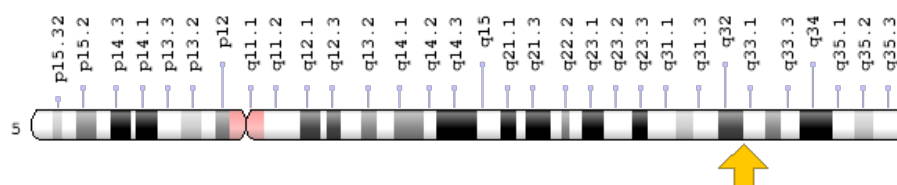
The protein produced from the *ETV6-PDGFRB* fusion gene (as well as other *PDGFRB* fusion genes) functions differently than the proteins normally produced from the individual genes. The ETV6/PDGFR $\beta$  fusion protein does not require ligand binding to be activated and cannot bind to DNA to turn off gene activity. As a result, signaling pathways are constantly turned on (constitutively activated) and gene activity is increased, which increases the proliferation and survival of cells.

When the *ETV6-PDGFRB* fusion gene mutation occurs in cells that develop into blood cells, the growth of eosinophils (and occasionally other white blood cells, such as neutrophils and mast cells) is poorly controlled, leading to *PDGFRB*-associated chronic eosinophilic leukemia. It is unclear why eosinophils are preferentially affected by this genetic change.

### Chromosomal Location

Cytogenetic Location: 5q32, which is the long (q) arm of chromosome 5 at position 32

Molecular Location: base pairs 150,113,839 to 150,155,884 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- beta-type platelet-derived growth factor receptor
- CD140 antigen-like family member B
- CD140B
- PDGF-R-beta
- PDGFR-1
- PDGFR-beta
- PDGFR1
- PGFRB\_HUMAN
- platelet-derived growth factor receptor 1
- platelet-derived growth factor receptor beta
- platelet-derived growth factor receptor, beta polypeptide

## Additional Information & Resources

### Educational Resources

- Holland-Frei Cancer Medicine (sixth edition, 2003): Chromosomal Rearrangements  
<https://www.ncbi.nlm.nih.gov/books/NBK12538/#A1403>
- Holland-Frei Cancer Medicine (sixth edition, 2003): Growth Factor Receptors  
<https://www.ncbi.nlm.nih.gov/books/NBK13714/#A1392>
- Holland-Frei Cancer Medicine (sixth edition, 2003): PDGFRB as a Therapeutic Target  
<https://www.ncbi.nlm.nih.gov/books/NBK12453/#A13684>

### GeneReviews

- Primary Familial Brain Calcification  
<https://www.ncbi.nlm.nih.gov/books/NBK1421>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28PDGFRB%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

### OMIM

- PLATELET-DERIVED GROWTH FACTOR RECEPTOR, BETA  
<http://omim.org/entry/173410>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
<http://atlasgeneticsoncology.org/Genes/PDGFRBID21ch5q32.html>
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=PDGFRB%5Bgene%5D>
- HGNC Gene Family: CD molecules  
<http://www.genenames.org/cgi-bin/genefamilies/set/471>
- HGNC Gene Family: I-set domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/593>
- HGNC Gene Family: Immunoglobulin like domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/594>
- HGNC Gene Family: Receptor Tyrosine Kinases  
<http://www.genenames.org/cgi-bin/genefamilies/set/321>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=8804](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8804)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/5159>
- UniProt  
<http://www.uniprot.org/uniprot/P09619>

## **Sources for This Summary**

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